10th European Conference on Rare Diseases & Orphan Products 2020

PhD Study Aim
To explore the relationship between Quality of Life and Social Support for adults living with rare genetic skin conditions.

Participants
Thirty-one adults with diagnosed rare genetic skin and connective tissues conditions including Ehlers-Danlos Syndrome (EDS) and Epidermolysis Bullosa (EB).

Stories are not just for bed time

Outcomes
- Some participants reported that they felt stigmatised, a sense of social isolation and experienced difficulties in managing the emotional impact of their condition.
- Analysis indicated a positive correlation between QoL and Social Support.
- The majority of the emotional support was provided by the family.

Quality of Life Measures
QoL measures were compared. All included physical, psychological and social dimensions. Most rare disease research is focused on the physical dimensions.

<table>
<thead>
<tr>
<th>QUALITY OF LIFE MEASURES</th>
<th>DIMENSIONS</th>
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<tbody>
<tr>
<td>Children’s Dermatology Life Quality Index</td>
<td>Physical</td>
</tr>
<tr>
<td>Dermatology Life Quality Index</td>
<td>Yes</td>
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<tr>
<td>Euro-Quality of Life 5 Dimension Questionnaire</td>
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<tr>
<td>Family Dermatology Life Quality Index</td>
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<td>Quality of Life in Epidermolysis Bullosa</td>
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<tr>
<td>Short Form 36</td>
<td>Yes</td>
</tr>
<tr>
<td>SKINDEX</td>
<td>Yes</td>
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<tr>
<td>World Health Organisation Quality of Life Questionnaire</td>
<td>Yes</td>
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</tbody>
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Impact of Diagnosis
“Once you get a diagnosis, that is more or less where the support stops”. “It affected me massively. I just went completely off the rails….I just completely shut down”.

Conclusion
Most rare disease research is focused on the physical aspects of conditions. Current world issues has taught us that adopting a holistic bio-psycho-social approach has to be a priority to effectively support the Rare Disease Community.

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